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PREVALENCE OF FABRY DISEASE IN KOREAN MALE PATIENTS WITH LEFT VENTRICULAR HYPERTROPHY

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Background: Fabry disease is an X-linked recessive disorder caused by a deficiency of lysosomal enzyme alpha-galactosidase A (α-Gal A). Previous studies have demonstrated that many patients with Fabry disease have been identified among male patients with left ventricular hypertrophy or hypertrophic cardiomyopathy. Recently, a variant form of Fabry disease was identified with manifestations primarily limited to the heart. The purpose of the present study was to define the incidence of Fabry disease among Korean male patients with left ventricular hypertrophy.

Methods: We prospectively studied male patients who were seen at the 10 Korean University Hospitals from January 2004 to December 2008. A total of 988 consecutive, unselected Korean male patients with left ventricular hypertrophy on echocardiography were screened for Fabry disease by measuring their plasma α-Gal A activities. The criterion for the diagnosis of left ventricular hypertrophy was a maximum left ventricular wall thickness $\geq$13 mm. Clinical manifestations in patients with low plasma α-Gal A activities were assessed and the α-Gal A gene mutations were screened.

Results: Nine (0.9%) of the 988 male patients with left ventricular hypertrophy had no or low plasma α-Gal A activities (α-Gal A activity $\leq$2.0 nmoles/hr/ml). Of these 9 patients, ranging in age from 24 to 85 years, 4 had hypertension and 3 had renal insufficiency. Left ventricular hypertrophy was concentric in 6 patients and asymmetric in 3 patients. Five of the 9 patients analyzed had α-Gal A gene mutations. α-GAL A activity in the remaining 979 patients was 9.4 $\pm$ 4.8 nmol/h/mL.

Conclusion: We detected Fabry disease in 0.9% of unselected Korean male patients with left ventricular hypertrophy. Although the prevalence of Fabry disease was low in our study, routine screening of male patients with left ventricular hypertrophy would enable earlier identification of many other affected relatives in their families who might benefit from specific clinical treatment.